Mixed Histocytosis Manifesting as Suprasellar Mass with Aortic Involvement

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Learning Objectives

- Define histiocytoses, LCH, N-LCH, and mixed histiocytosis.
- Explain the spectrum of L-type histiocytosis.
- Discuss difference in organ system involvement.
- Examine difficulty in diagnosis.
- Discuss importance of imaging, cytology, and clinical suspicion in diagnosis.

Introduction

Histiocytoses are group of disorders affecting myeloid progenitor cells. The etiopathogenesis of histiocytoses are largely unknown, but BRAF-V600E mutation is frequently implicated in the pathogenesis of these conditions.

Histiocytoses are grouped into L-C-M-R-H classification presenting with various symptomology, cytology, and prognosis dependent on category.

- The L-group includes Langerhan, Non-Langerhan, and Mixed histiocytosis.
- Langerhan Cell Histiocytosis (LCH) incidence is one case per million adults.
- Non-Langerhan Cell Histiocytosis (N-LCH) have only 500 documented cases and Mixed Histiocytosis only around 100 cases (2-3 others).

Histiocytoses clinical manifestations vary depending on organ involvement.

- LCH has been known to affect long bones, skin, lymph nodes, spleen, and CNS.
- N-LCH can affect long bones, axial inflammatory, large blood vessels, heart, CNS, and skin and pituitary gland (2).

Diagnosis is made with tissue biopsy with cytology, clinical manifestations, and imaging showing multisystem involvement.

- Cytology testing include CD1a, CD68, CD123, S100, BRAFV600E, LCH, and N-LCH carry a poor prognosis, which worsens with CV and CNS involvement.
- Treatment for LCH, N-LCH, and Mixed Histiocytosis vary so differentiation is important.
- LCH treatment is centered off radiation, resection, and targeted chemotherapy.
- N-LCH has no standardized treatment but trials show benefit with adding PEG to unlike LCH.

Case Presentation

In August, a 42-year-old white female with a past medical history of migraines, depression, and fibromyalgia initially presented to Henry Ford Macomb Hospital after a syncopal event. The event occurred after having sutures removed one week status-post tonsillectomy surgery and was transported to the ER immediately for evaluation.

Upon arrival, the patient was feeling light-headed with associated headaches, visual changes, diaphoresis and fatigue which she had not experienced in the past. The patient denied symptoms of fever, shortness of breath, nausea, vomiting, focal weakness, or loss of sensation. The patient family history and social history were non-contributory. Her past surgical history was significant for tonsillectomy release, cholecystectomy, sleeve gastrectomy, and umbilical hernial repair. The patient has no known drug allergies. The patient medications included cetirizine, cyproheptadine, docusate, lactulose, doxapram, ativan, omeprazole, sertraline, tizzadine, and tramadol. Physical exam was notable for an anxious woman in mild distress, appraising pale, no obvious signs of trauma, normal heart and lung sounds, no abdominal tenderness, and no focal neurologic deficits. The patients vitals revealed a blood pressure of 71/47mmHg, HR 84bpm, 97.2°F (temporarily, RR 16bpm, and SpO2 96% on room air.

Initially the event was presumed to be vasovagal in nature but a syncopal workup ensued with an EKG, CTA CT head and neck, and blood work. EKG was abnormal with a prolonged QT interval of 612 which was not previously diagnosed. CTA with pulmonary embolism protocol incidentally found inflammation of the aorta and left subclavian vessel with mural thickening of descending aorta. CTA also showed stenosis of the celiac, superior mesenteric, and renal arteries. This finding was suspicion of Takayasu arteritis.

Takayasu arteritis. CTA head and neck revealed 14mm hypointense mass with optic chiasm involvement—the patient denied visual impairment. The patient was discharged with plans to follow up with her rheumatologist and neurosurgery for evaluation of her hypothalamic mass. In December 2018, due to worsening of vision, an MRI imaging of the patients brain was obtained showing enlargement of the hypothalamic mass, measuring 21.3mm with associated intermittent horizontal diplopia occurring daily with continued fatigue, fever, and cold intolerance. Within a month, the patient had a right craniotomy performed with partial tumor resection and was complicated by post-operative cerebral edema and uncal herniation. Patient was given Decadron and intubated for a week to preserve the airway. The patient subsequently developed diabetes insipidus which was treated with desmopressin. Endocrinology diagnosed panhypopituitarism secondary to resection and hormone replacement therapy was initiated. The patient is still being treated for her condition.

Clinical Imaging

Figure 1: (above left) MRI of head 21.6mm suprasellar mass which appears to be centered on the pituitary stalk neural hypophysis Mass effect upon the hypothalamic and optic chiasm.

Figure 2: (above right) MR-Angiography of Chest/Abdomen Multiple findings consistent with Takayasu Arteritis Diffuse mural thickening of the aortic arch + descendng thoracic aorta—thickness (6mm) Moderate stenoses of the proximal portions of the left subclavian artery and common carotids.

Laboratory Values

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<thead>
<tr>
<th>Table 2: Laboratory Results</th>
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<tbody>
<tr>
<td>Hematocrit (Hct)</td>
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<tr>
<td>38.0 – 45.0</td>
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<td>White Blood Cell (WBC)</td>
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<td>Platelets (PLT)</td>
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References

- Histiocytoses: an overview. The different and hard to recognize presentations, such as this case makes classification of histiocytoses difficult as well. The subclassifications within the L-group of LCH, N-LCH, and Mixed histiocytosis are nebulous. Cases of these conditions are exceptionally rare which make the classification system difficult to effectively distinguish. The classification of these groups are primarily cytologic, but is this the best way to classify these conditions clinically? These processes pathogenesis are widely unknown, but are known as be error in myeloid progenitors differentiation—presumptively at different locations. This could provide context to why the tumors cell markers vary even amongst the same class. The biopsy for this case revealed CD-1a positive yet its CD-163/CD-68 positivity technically classified it “Non-Langerhan Cell” Different lesions, such as this patients, doesn’t fit distinctly into any of the three as aforementioned categories which make the current schema seem relatively ineffective. This patients clinical presentation with vascular involvement was more consistent with N-LCH, however the biopsy was inconclusive between LCH and N-LCH—making it more consistent with mixed histiocytosis. Since cytokoly can be inconclusive especially in light of clinical findings, it makes the L-group of histiocytosis seem to be more of a spectrum of disease rather than distinct categories.

Conclusion

- Histiocytoses present as a rare spectrum of disease with different presenting symptoms.
- Differences in diagnosis depends primarily on cytology and organ system involvement.
- Treatment protocols vary for different histiocytosis although guidelines are not well established.
- Patients that present under these rare condition should consider enrolling in clinical trials.

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